VIII - ABSTRACT

The present invention concerns the development of a quantitative method for the molecular diagnosis of autosomal recessive spinal muscular atrophy (SMA) by measuring the amount of cytosolic mRNA from human muscle cells. Both the procedure using radioactive material and the Enzyme-Linked Immunosorbent Assay (ELISA) nonradioactive method were developed using ^{32}P-dCTP labeled and biotinylated nucleotide probes. The results obtained demonstrate that the measurement of mRNA could be used as a quantitative method for the molecular diagnosis of SMA. There was a perfect concordance of the results obtained between the procedure using radioactive material, the ELISA method and the single strand conformation polymorphism (SSCP) analysis regarding the negative and positive SMA samples. The methods developed in this study may be applicable to the diagnosis (detection of homozygous and heterozygous deletions in exons 7 and 8 of the SMN gene) and the control of mRNA concentrations in the future gene therapy of patients with SMA.